

# **PARENT FACT SHEET**

## **DISORDER**

### **Medium-chain acyl-CoA dehydrogenase deficiency (MCAD)**

#### **CAUSE**

MCAD occurs when an enzyme called “medium chain acyl-CoA dehydrogenase” is either missing or not working properly. This enzyme’s job is to break down certain fats in the food we eat into energy. It also breaks down fat already stored in the body. Energy from fat keeps us going whenever our bodies run low of their main source of energy, a type of sugar called glucose. Our bodies rely on fat when we don’t eat for a period of time.

#### **IF NOT TREATED**

When the MCAD enzyme is missing or not working, the body cannot use certain types of fat for energy and must rely solely on glucose. Although glucose is a good source of energy, there is a limited amount available. Once the glucose has been used up, the body tries to use fat without success. This leads to low blood sugar, called hypoglycemia, and to the build of harmful substances in the blood. This is called a metabolic crisis. Metabolic crisis can cause permanent brain damage and even death.

#### **TREATMENT OPTIONS**

Your child will need to be under the care of a metabolic specialist and dietician. Treatment is usually needed throughout life.

- Your child needs to avoid going a long time without food. This is to avoid a metabolic crisis. These children should not go more than 4 to 6 hours without food and some may require more frequent feedings. It is important that these children be fed in the night – meaning you will need to wake them up to eat if they do not wake up on their own – and even if they are not hungry.
- Your metabolic specialist and dietician will design a low-fat, high carbohydrate diet for your child. This may include a special formula when your child is an infant. No changes in diet can happen without first getting permission from the metabolic specialist and dietician.
- Some children may benefit from taking L-carnitine. This is a safe and natural substance that helps the body make energy. The metabolic specialist will decide if your child can benefit from this treatment. A prescription for this is required.
- Contact your child’s doctor immediately at the start of any illness. Children with MCAD need to be treated in a hospital to prevent serious health problems.

#### **IF TREATED**

With prompt and careful treatment, children with MCAD usually live health lives with typical growth and development. The goal of treatment is to prevent long-term problems. However, children who have repeated metabolic crises may have life-long learning disabilities, uncontrolled movements in their muscles and tendons, or other effects.

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